



TGM5 gene

transglutaminase 5

Normal Function

The *TGM5* gene provides instructions for making an enzyme called transglutaminase 5. This enzyme is found in many of the body's tissues, although it seems to play a particularly important role in the outer layer of skin (the epidermis). In the epidermis, transglutaminase 5 is involved in the formation of the cornified cell envelope, which is a structure that surrounds cells and helps the skin form a protective barrier between the body and its environment. Specifically, transglutaminase 5 forms strong bonds, called cross-links, between the structural proteins that make up the cornified cell envelope. This cross-linking provides strength and stability to the epidermis.

Health Conditions Related to Genetic Changes

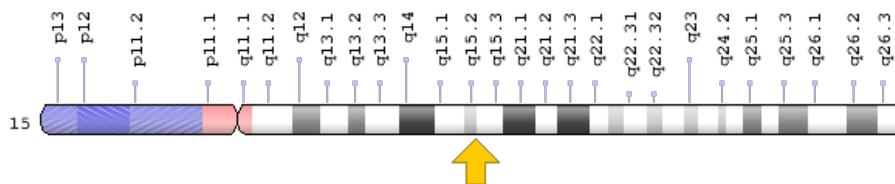
acral peeling skin syndrome

At least 22 mutations in the *TGM5* gene have been found to cause acral peeling skin syndrome. This condition is characterized by painless peeling of the top layer of skin that is most apparent on the hands and feet but can also affect the arms and legs. Most of the mutations change single protein building blocks (amino acids) in transglutaminase 5, including the most common mutation in people of European ancestry, which replaces the amino acid glycine with the amino acid cysteine at position 113 (written as Gly113Cys or G113C). *TGM5* gene mutations reduce the amount of transglutaminase 5 that is produced or prevent cells from making any of this enzyme. A shortage of transglutaminase 5 impairs protein cross-linking, which weakens the cornified cell envelope and allows the outermost cells of the epidermis to separate easily from the underlying skin and peel off. This peeling is most noticeable on the hands and feet probably because those areas tend to be heavily exposed to moisture and friction.

Chromosomal Location

Cytogenetic Location: 15q15.2, which is the long (q) arm of chromosome 15 at position 15.2

Molecular Location: base pairs 43,232,595 to 43,266,857 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- protein-glutamine gamma-glutamyltransferase 5
- protein-glutamine gamma-glutamyltransferase 5 isoform 1
- protein-glutamine gamma-glutamyltransferase 5 isoform 2
- TG(X)
- TGase-5
- TGase X
- TGASE5
- TGASEX
- TGM5_HUMAN
- TGM6
- TGMX
- TGX
- transglutaminase-5
- transglutaminase V
- transglutaminase X

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Epidermal Cells Form a Multilayered Waterproof Barrier
<https://www.ncbi.nlm.nih.gov/books/NBK26865/#A4082>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TGM5%5BTIAB%5D%29+OR+%28transglutaminase+5%5BTIAB%5D%29+OR+%28TG5%5BTIAB%5D%29%29+OR+%28%28TGASE5%5BTIAB%5D%29+OR+%28transglutaminase-5%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- TRANSGlutaminase 5
<http://omim.org/entry/603805>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TGM5.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TGM5%5Bgene%5D>
- HGNC Gene Family: Transglutaminases
<http://www.genenames.org/cgi-bin/genefamilies/set/773>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11781
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9333>
- UniProt
<http://www.uniprot.org/uniprot/O43548>

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